Newborn Screening

New Initiatives and Building on Lessons Learned

March 29, 2017

South Carolina Department of Health and Environmental Control

Healthy People. Healthy Communities.

South Carolina Hospital Association

continuing medical education and continuing nursing education accreditation and credit

PALMETTO HEALTH

UNIVERSITY OF SOUTH CAROLINA School of Medicine

SCHA South Carolina Hospital Association

continuing medical education accreditation statements

- This activity has been planned and implemented in accordance with the Essential Areas and policies of the Accreditation Council for Continuing Medical Education (ACCME) through the joint sponsorship of the University Of South Carolina School Of Medicine – Palmetto Health CME Organization and the South Carolina Hospital Association. The University of South Carolina School of Medicine – Palmetto Health CME Organization is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.
- The University of South Carolina School of Medicine – Palmetto Health CME Organization designates the live activity, South Carolina Surgical Quality Collaborative Annual Meeting, for a maximum of *1.0 AMA PRA Category 1 Credits™*. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

continuing nursing education accreditation statements

- The South Carolina Hospital Association is an approved provider of continuing nursing education by the South Carolina Nurses Association, an accredited approver by the American Nurses Credentialing Center's Commission on Accreditation.

- Participants who attend the conference and complete an evaluation will earn 1.0 contact hours.

disclosure statements

Relevant financial relationships will be disclosed to participants at the conference. Faculty members are required to disclose off-label/investigative uses of commercial products/devices.

The activity planning committee members have disclosed that they have no relevant financial relationships. There is no commercial support for this activity.

continuing education

In order to receive continuing education credit hours, please sign in at the registration table and confirm your email address.

For those participants seeking continuing medical education, please sign in and sign out.

The evaluation will be emailed to you after the conference and you will receive a certificate upon completion. CME certificates will be mailed.

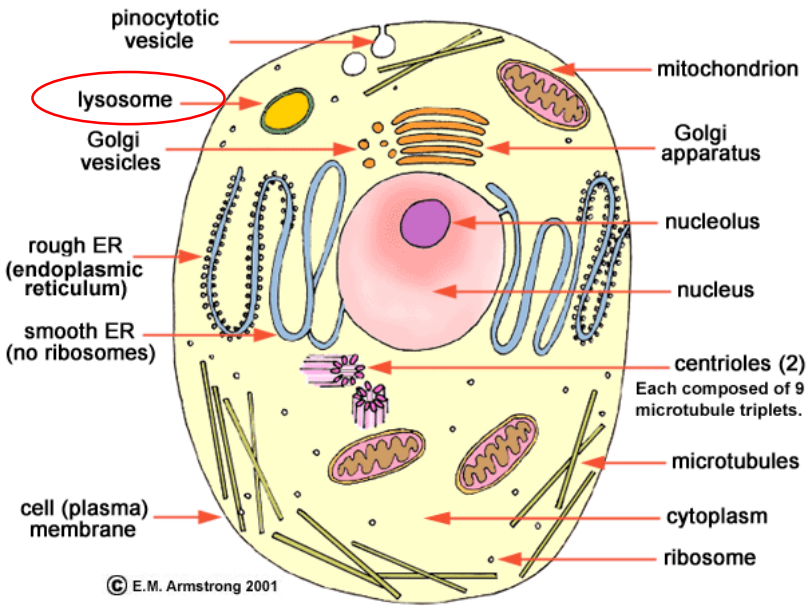
Thank you to the South Carolina Hospital Association for their continued support in helping us improve Newborn Screening processes and ensuring the best possible care for the babies of South Carolina.

South Carolina
Hospital Association

Lysosomal Storage Disorders

- Added to the Recommended Uniform Screening Panel (RUSP) Core Conditions in March 2015.
  - Published by USDHHS, Advisory Committee on Heritable Disorders in Newborns and Children
- Ad Hoc Advisory Committee Convened for Implementation
- Pompe Disease (Glycogen Storage Disease Type II)
- MPS-1 (Mucopolysaccharidosis Type 1)
- X-ALD (X-linked Adrenoleukodystrophy)

Errors in the Organelle



- The lysosome is the place of cellular digestion and recycling of proteins, carbohydrates and lipids.
- Disorder = lack of enzyme

Pompe Disease

Overview

- Accumulation of glycogen in the lysosome due to an absence of the enzyme α-glucosidase.
- The toxic glycogen build up damages organs and tissues, specifically smooth muscles like the heart.

Clinical Features and Prevalence

- Presents at 1-2 months of life
- Profound hypotonia and hyporeflexia
- Cardiomegaly
- Enlarged Tongue
- Hearing Loss
- Normal Cerebral Development
- **FATAL** by 1 year of life if untreated.
- 1:28,000-40,000, 2-3 cases in SC/yr

Pompe Disease

Diagnosis

- Elevated CK, LFTs
- Echocardiogram
- Biochemical
  - α-glucosidase enzyme deficiency or malfunction

Treatment

- **Enzyme replacement therapy** (**ERT**) is available and is intended to replace the deficient or absent **acid alpha-glucosidase** (GAA) enzyme.

Mucopolysaccharidosis Type 1

Overview

- MPS 1 is a rare genetic disorder that affects many body systems and leads to organ damage.
- It is caused by a mutation in the gene that makes the enzyme alpha-L-iduronidase .
- Forms are Hurler, Hurler-Scheie and Scheie syndromes.

MPS 1
Clinical Features

- Abnormal Bone formation
- Joint stiffness
- Cardiovascular disease
- Course facial features
- Carpal Tunnel
- Hearing loss
- Obstructive airway disease
- Enlarged liver and spleen
- Hydrocephalus
- Corneal clouding
- Delayed mental development
- Spinal Cord Compression

MPS 1

- Diagnosis
- Elevated urine GAGs (glycosaminoglycans)
- Deficiency or absence of alpha-L-iduronidase in blood or skin cells
- DNA testing

- Treatment
- No cure, but bone marrow transplant and/or ERT can help.
- **Laronidase** therapy has improved walking capacity and pulmonary function.
- Idursulfase is a purified form of human iduronate-2-sulfatase, a lysosomal enzyme.

X-linked Adrenoleukodystrophy

Overview

- Adrenoleukodystrophy (**X-ALD**) is a serious progressive, genetic disorder that affects the adrenal glands, the spinal cord and the white matter of the nervous system.
- It was first recognized in 1923 and has been known as Schilder's disease and sudanophilic leukodystrophy.

X-ALD

Clinical Features

- Symptoms range from a progressive disease of the spinal cord in men and women:
- Adrenomyeloneuropathy (AMN) to a fatal brain disease in boys and men (cerebral **ALD**).
- **ALD** is caused by a genetic defect (mutation) in the *ABCD1* gene.

X-ALD
Diagnosis

- High blood levels of very long-chain fatty acids (VLCFA)
- Genetic testing to identify defects or mutations that cause ALD
- MRI
- Vision Screening
- Skin Biopsy and Fibroblast Cell Culture

X-ALD

Treatment

- No cure, but **Lorenzo's oil** and **stem cell transplantation**, using either umbilical cord stem cells or bone marrow stem cells are helpful.
- Adrenal Insufficiency Treatment
- Corticosteroid replacement therapy
- Medications
- Physical Therapy
- Genetic Counseling

Lysosomal Storage Disorders
Laboratory needs

SAMPLE:

- Pompe Disease & MPS-I
  - 1 additional 3.2 mm punch from the dried blood spot (DBS)
- X-ALD
  - No additional 3.2 mm punch needed; can be analyzed from the current punch for Tandem Mass Spectrometry (MSMS) testing for Amino Acids, Acylcarnitines, and Succinylacetone (AA/AC/SA)

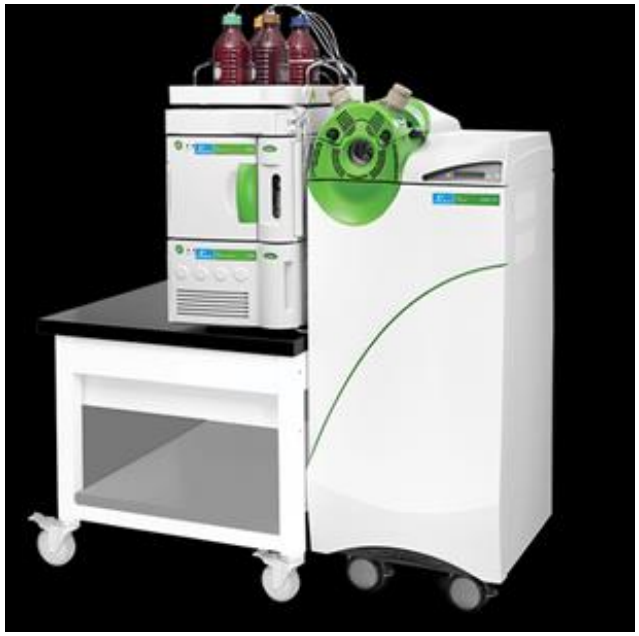
Lysosomal Storage Disorders
Laboratory needs, cont'd

INSTRUMENTATION:

Pompe Disease, MPS-I, & X-ALD

- 1 additional MSMS analyzer will be acquired for the addition of Pompe & MPS-I

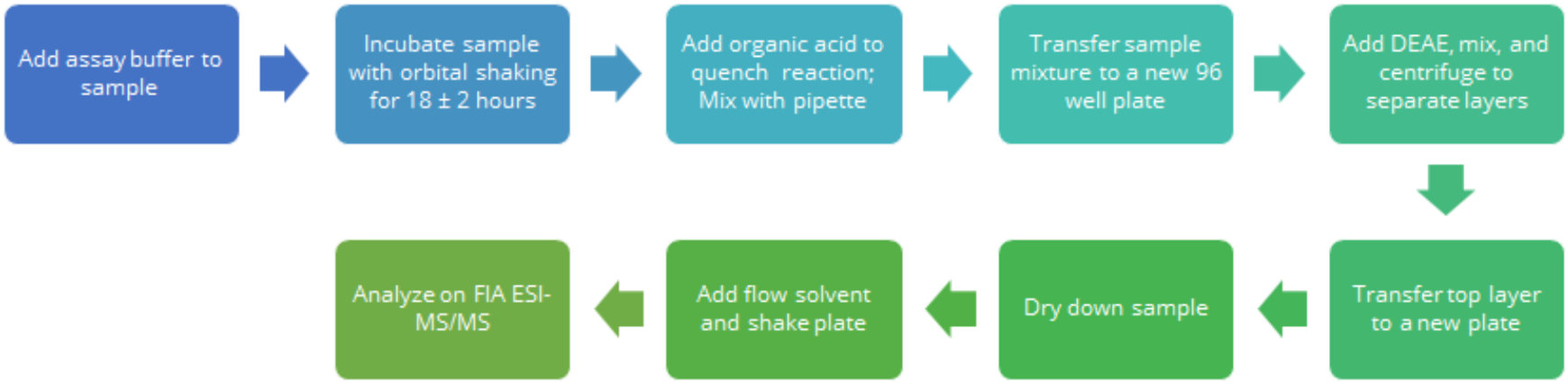
- 2 identical instrument platforms will be acquired for the current MSMS testing, along with the addition of X-ALD



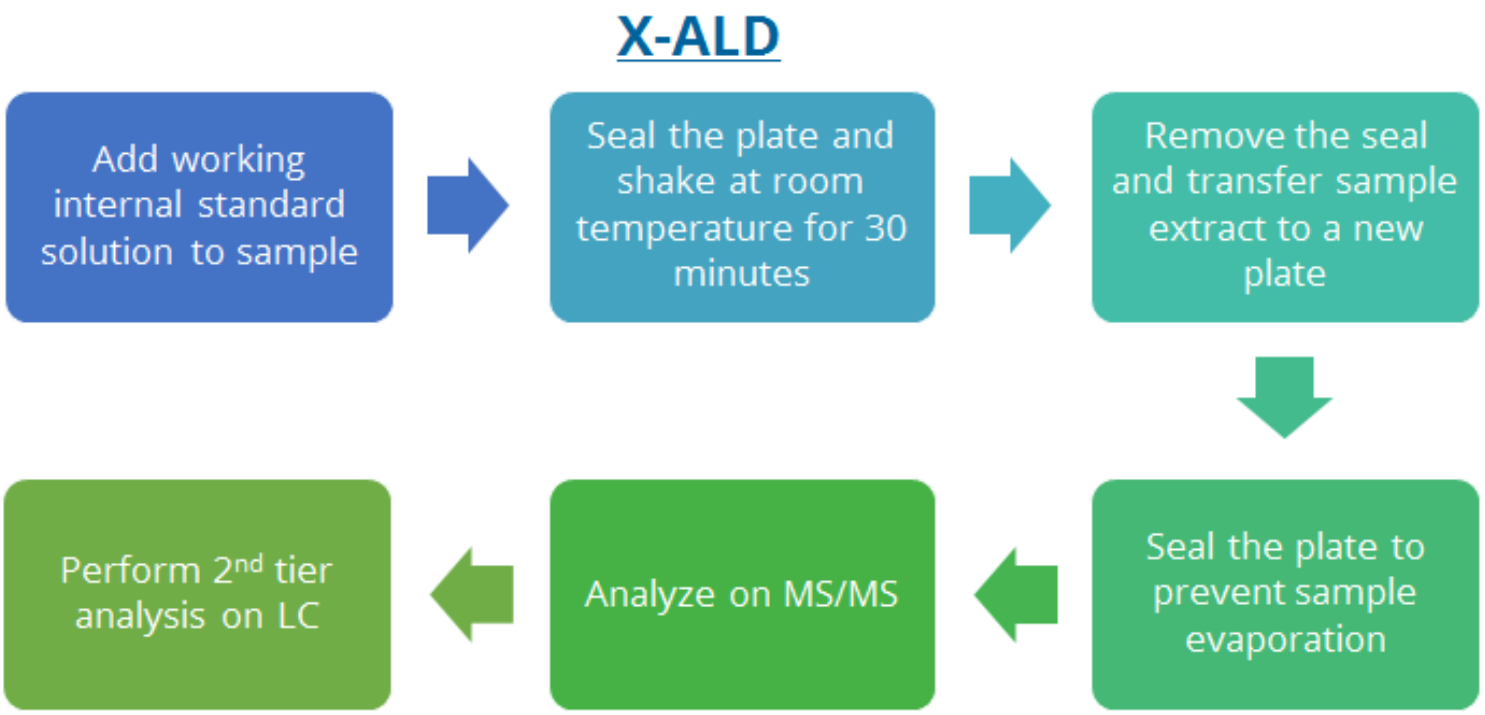
http://www.perkinelmer.com/lab-solutions/resources/images_for_resize//QSight-station_500x500.png

Lysosomal Storage Disorders
Laboratory workflow

Pompe Disease & MPS-I



Lysosomal Storage Disorders
Laboratory workflow



Building on Lessons Learned On Time Every Time Initiative

- In 2014, a collaboration with SCHA and the newborn screening program began providing monthly QI measurements to SC hospitals. These measures were to increase the timeliness in which specimen were received to the DHEC Bureau of Laboratories.
- The intent was to reach the national CLSI goals for time critical conditions being identified by 5 days of life and non-critical conditions being identified by 7 days of life.
- In 2017, a report card was distributed to all SC hospitals to show achievement of quality improvement measures and in newborn screening.

Building on Lessons Learned: Overview of QI Measures

Unsatisfactory Specimens

A measure of the percent of unacceptable dried blood spot specimens due to improper collection or transport.

** SC statewide goal is 2%**

Time between birth and collection

A measure to ensure that a specimen is collected between 24-48 hours of life

** SC statewide goal is 1-2 days**

Building on Lessons Learned: Overview of QI measures

Time between collection and postmark

A measure to ensure that a specimen is postmarked and mailed in a timely manner after collection. A postmark is considered as the date that is stamped by the mailing facility and should be performed after the specimen is collected and prior to being mailed.

** SC statewide goal is 1 day**

Building on Lessons Learned Overview of QI Measures

Time between postmark and DHEC receipt

A measure to ensure timeliness between mailing of a specimen and receipt at the DHEC newborn screening lab.

** SC statewide goal is 0% >5 days after collection**

QI Measures
2016 Averages for Timeliness

Time between birth and collection

1.42 days

Time between collection and DHEC receipt

2.42 days

Newborn Screening Specimen Transport Protocol

- The South Carolina Department of Health and Environmental Control (DHEC) is pleased to announce the start of a new transportation protocol for newborn screening specimens.

- Beginning as early as April 1st, FedEx will pick up newborn screening dried bloodspot filter cards from our submitters and deliver them via FedEx Priority Overnight® service to the DHEC Bureau of Laboratories located at 8231 Parklane Road in Columbia, SC **at no additional cost to the submitter**. The delivery service is part of the new fee structure for newborn screening testing.

- As the new protocol is rolled out, you will receive a separate e-mail with a login username and password for access to FedEx online shipping website. All newborn screening shipping will be processed online. The website is user-friendly. However, FedEx technical support is available to your site by phone, if necessary.

Newborn Screening Specimen Transport Highlights

- *New NBS fee in effect on April 1, 2017 is $127*
- Includes new specimen transport protocol
- Partnership between DHEC and FedEx
- *No additional cost to the submitter*
- All shipping processing done online
- Submit contact information to:

washints@dhec.sc.gov

adairoo@dhec.sc.gov

Shared best practices for Timeliness

Trident Medical Center
Nancy Quire, R.N.
Line Service Director

Nancy.Quire@hcahealthcare.com

South Carolina Department of Health and Environmental Control
Healthy People. Healthy Communities.

Newborn Screening Process Trident Health

TRIDENT HEALTH

Newborn Screening Process

Work flow process for Department Leaders:

- Train each employee to ensure that the Newborn Screening Form is completely filled out accurately.
- Train each employee to ensure blood is collected correctly. Each circle should be completely filled out.
- On Monday the department manager prints the FedEx labels to send samples Monday - Sunday
- Women's and Children's staff member delivers samples to mailroom before 4:00 p.m. each day.
- Saturday morning FedEx is alerted to pick up envelope shipments directly from the floor.

Continuous monitoring of the timing of the shipments and continually investigating improvement strategies.

Newborn Screening Process

Workflow process for Women's and Children's staff:

- Newborn Screen form is completed by the nursing staff:
  - Allow specimen to dry completely
  - After dry, the specimens are placed in DHEC Bureau of Laboratories envelope and sealed
  - Dated forms are placed in a dated brown envelope
  - DHEC envelope is placed in a FedEx Express mailer with printed shipping information
  - Monday-Friday the FedEx envelope is hand carried to the FedEx drop box for pick-up before 4:00 p.m.
  - Saturday- Sunday specimens are prepared by night shift nursery charge nurse and is picked up by courier on the unit

Building on Lessons Learned: First Time Every Time initiative

In 2016, a collaboration with SCHA and the Newborn Screening Program hosted 6 regional training workshops with a "Train the Trainer" focus. This focus was to assist with decreasing unsatisfactory specimen submission and the need for a repeat specimen to be collected. Participants received training on correct dried blood spot collection procedures with a "hands on" approach.

77% of all SC hospitals participated by sending their nursing and lab staff to either Coastal Carolina, McLeod Regional Medical Center, Medical University of South Carolina, Palmetto Health Richland, or St. Francis Eastside.

Building on Lessons Learned Unsatisfactory Specimens

2016 Annual Unsatisfactory Rate **3.5%**

Percentage of unsatisfactory specimens in 2016 that *did not* get repeated

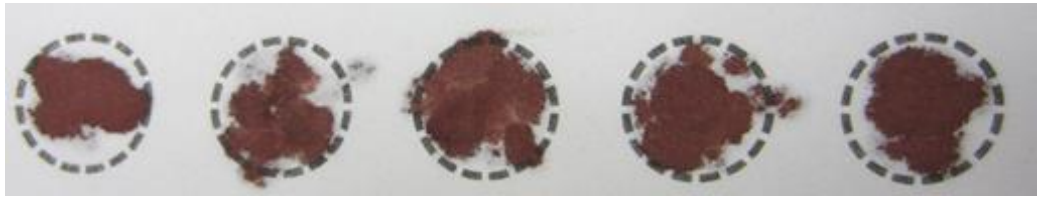
0.6%

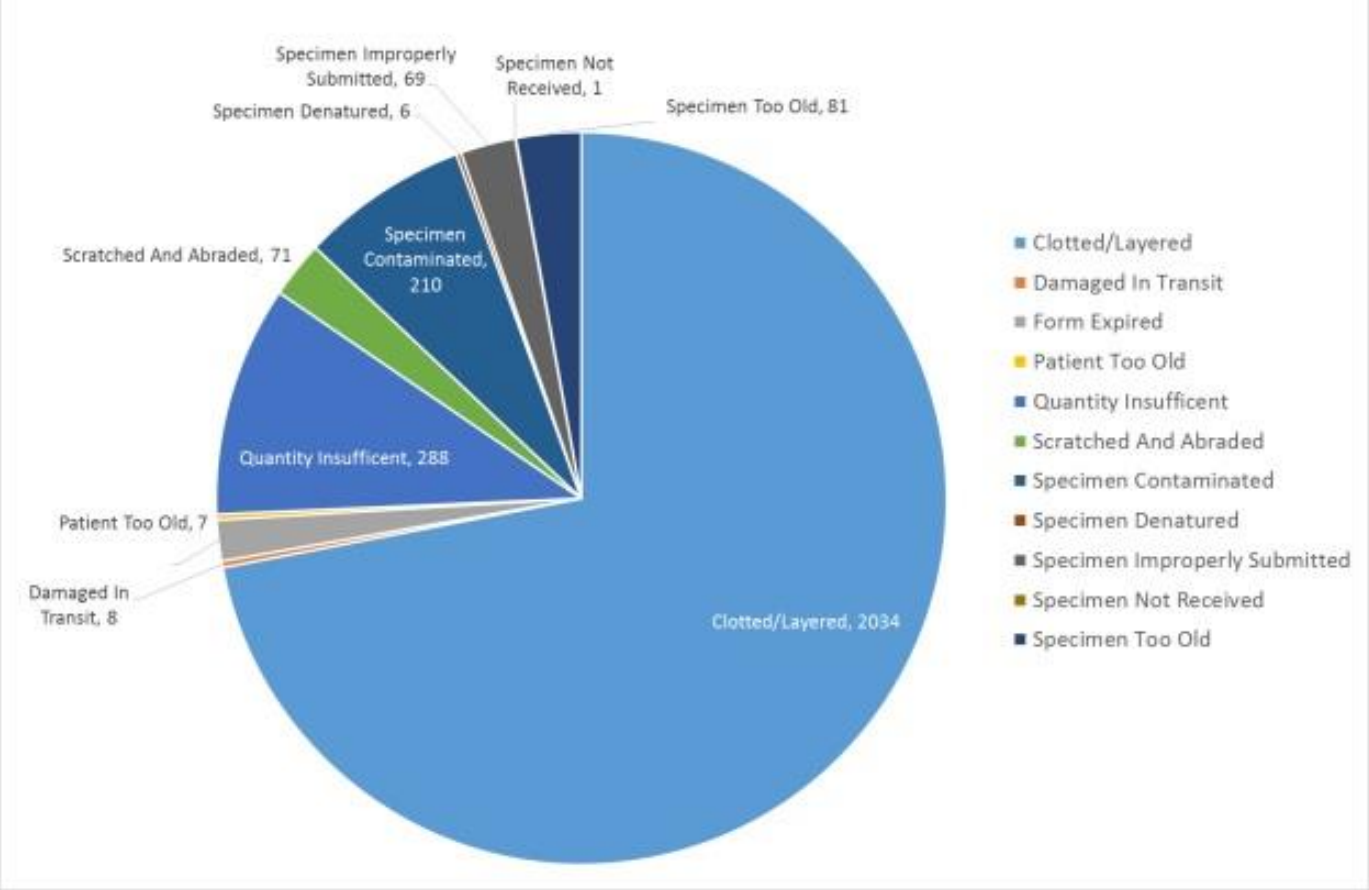
Percentages calculated from first samples collected

Building on Lessons Learned Unsatisfactory Specimens

- Most common Unsatisfactory Reason in 2016 is:

SPECIMEN IS CLOTTED OR LAYERED

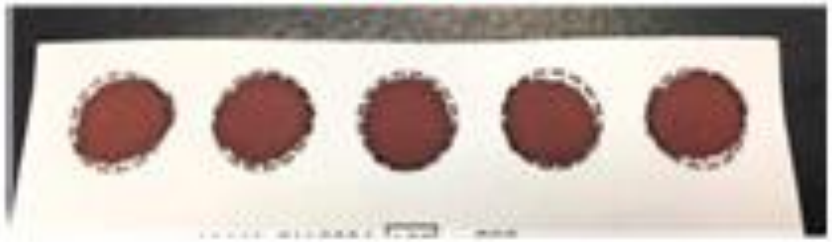



Specimen Improperly Submitted, 69
Specimen Not Received, 1
Specimen Denatured, 6
Specimen Too Old, 81
Scratched And Abraded, 71
Specimen Contaminated, 210
Quantity Insufficent, 288
Patient Too Old, 7
Damaged In Transit, 8
Clotted/Layered, 2034
Clotted/Layered
Damaged In Transit
Form Expired
Patient Too Old
Quantity Insufficent
Scratched And Abraded
Specimen Contaminated
Specimen Denatured
Specimen Improperly Submitted
Specimen Not Received
Specimen Too Old

SATISFACTORY Specimen

Front of Card

Back of Card



UNSATISFACTORY Specimen

Front of Card

Back of Card

Examples of UNSATISFACTORY Specimens

Quantity Not Sufficient (some tests can be performed)

Some layering and one spot is clotted

Clotted, Layered, and Abraded

Abraded

Building on Lessons Learned Unsatisfactory Specimens

Clotted and Layered-

What does this mean?

- Multiple drops of blood on the same circle on the NBS form
- Blood applied to both sides of the NBS form

How do you store your NBS forms?

- NBS forms should be stored vertically/upright on their side in the original packing

How shouldn't you store your NBS forms?

- Do not allow forms to compress by stacking/flat.
- Do not allow forms to be scratched or placed under objects.

Shared Best Practices to Improve Unsatisfactory Specimens

Tidelands Georgetown Memorial Hospital:
Debbie Hockensmith
Lab Director

Tidelands Health Georgetown Memorial Newborn Metabolic Screening

Process for Improvement in Collection and Submission

Initial State

- Primarily RN collected specimens, few from Lab Phlebotomy
- Many rejections from both RN and Lab Phlebotomy

Ideal State

- Eliminate all unsatisfactory reports caused by collection errors
- Prevent submission of improperly collected specimens from our facility
- Avoid any late submissions
- Do what is best for the newborn!

Tidelands Health Georgetown Memorial Newborn Metabolic Screening

Process for Improvement in Collection and Submission
Target (Real) State

- Collaborative decisions between Nursery and Laboratory
  - Nursery staff supports collections at suitable times for laboratory personnel
  - Lab-only collections will be done by exacting protocol
  - Lab will train technical staff to review and to process specimens for send out
  - Rejection will be done by technical staff as necessary before newborn discharged
- Robust retraining of all lab phlebotomists
  - Video training – found a training video from a reputable North Carolina site (Dept of Health)
  - Direct observation – soon super-collectors were identified and assisted in training
  - Ongoing competency includes reading the updated procedure, critical thinking skills and written testing
- Post-Analytical Auditing
  - Specimens are examined by lab technical staff immediately upon receipt directly after collection and again in 1 hour
  - Approved specimens are then dried for 3-4 hours in proper position and location
  - Internal log kept to identify each submission and ensure proper postmark applied
  - All specimens placed in lab/hospital courier designated area
  - Courier must mark how many specimens are picked up at each time and verify that log from lab matches courier pick up
  - Each envelope is clocked out and lab technical staff verifies the clocking
  - On weekends and most holidays, a backup system with USPS stamps is utilized with the clocking process as above

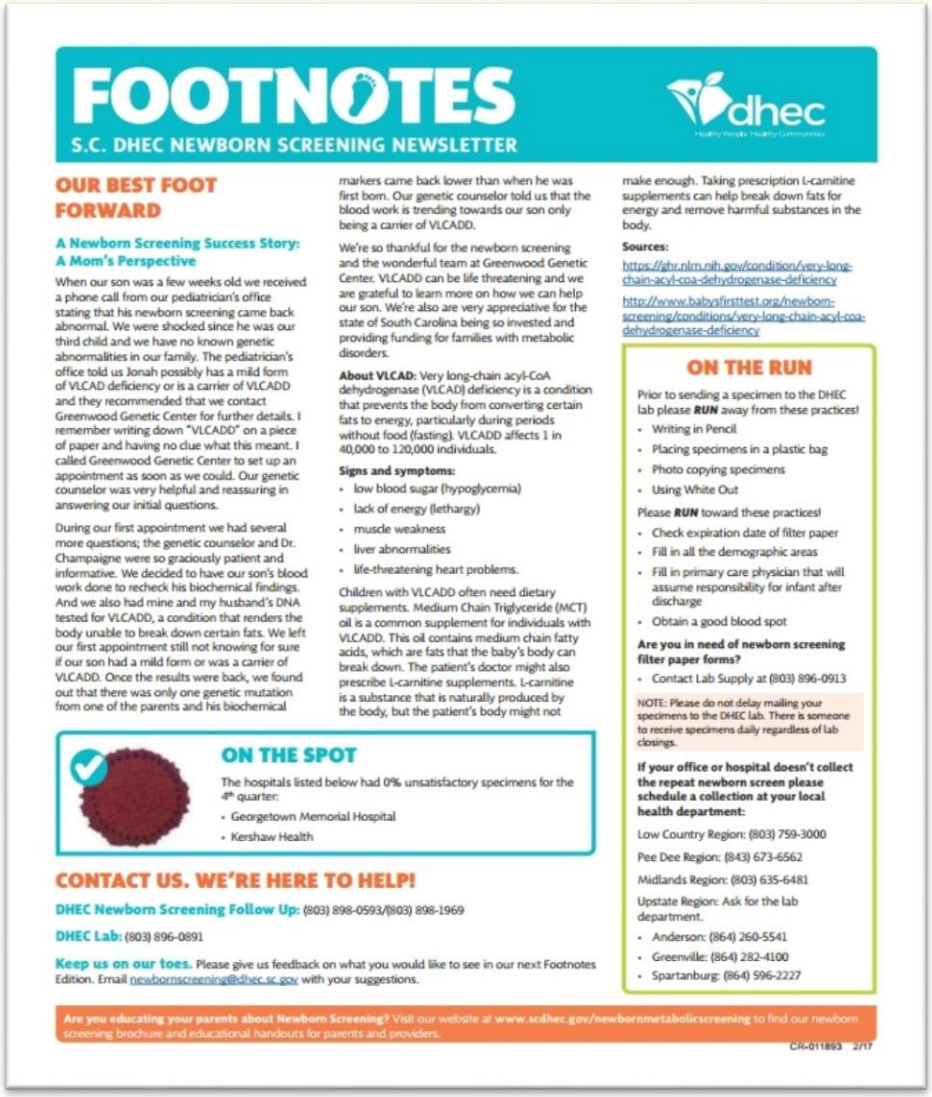
FOOTNOTES: Newborn Screening Newsletter

Our best foot forward: Update on newborn screening program progress, specific disorders, and patient stories.

On the run: Helpful tips and current practices provided by the newborn screening lab.

On the spot: Quarterly recognition of hospitals who have maintained 0% unsatisfactory rate.

Lab Closings: May 29th and July 4th



FOOTNOTES

S.C. DHEC NEWBORN SCREENING NEWSLETTER

OUR BEST FOOT FORWARD

A Newborn Screening Success Story: A Mom's Perspective

When our son was a few weeks old we received a phone call from our pediatrician's office stating that his newborn screening came back abnormal. We were shocked since he was our third child and we have no known genetic abnormalities in our family. The pediatrician's office told us Jonah possibly has a mild form of VLCAD deficiency or is a carrier of VLCADD and they recommended that we contact Greenwood Genetic Center for further details. I remember writing down "VLCADD" on a piece of paper and having no clue what this meant. I called Greenwood Genetic Center to set up an appointment as soon as we could. Our genetic counselor was very helpful and reassuring in answering our initial questions.

During our first appointment we had several more questions; the genetic counselor and Dr. Champaigne were so graciously patient and informative. We decided to have our son's blood work done to recheck his biochemical findings. And we also had mine and my husband's DNA tested for VLCADD, a condition that renders the body unable to break down certain fats. We left our first appointment still not knowing for sure if our son had a mild form or was a carrier of VLCADD. Once the results were back, we found out that there was only one genetic mutation from one of the parents and his biochemical markers came back lower than when he was first born. Our genetic counselor told us that the blood work is trending towards our son only being a carrier of VLCADD.

We're so thankful for the newborn screening and the wonderful team at Greenwood Genetic Center. VLCADD can be life threatening and we are grateful to learn more on how we can help our son. We're also are very appreciative for the state of South Carolina being so invested and providing funding for families with metabolic disorders.

About VLCAD: Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency is a condition that prevents the body from converting certain fats to energy, particularly during periods without food (fasting). VLCADD affects 1 in 40,000 to 120,000 individuals.

Signs and symptoms:

- low blood sugar (hypoglycemia)
- lack of energy (lethargy)
- muscle weakness
- liver abnormalities
- life-threatening heart problems.

Children with VLCADD often need dietary supplements. Medium Chain Triglyceride (MCT) oil is a common supplement for individuals with VLCADD. This oil contains medium chain fatty acids, which are fats that the baby's body can break down. The patient's doctor might also prescribe L-carnitine supplements. L-carnitine is a substance that is naturally produced by the body, but the patient's body might not make enough. Taking prescription L-carnitine supplements can help break down fats for energy and remove harmful substances in the body.

Sources:

https://ghr.nlm.nih.gov/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency

http://www.babysfirsttest.org/newborn-screening/conditions/very-long-chain-acyl-coa-dehydrogenase-deficiency

ON THE RUN

Prior to sending a specimen to the DHEC lab please ***RUN*** away from these practices!

- Writing in Pencil
- Placing specimens in a plastic bag
- Photo copying specimens
- Using White Out

Please ***RUN*** toward these practices!

- Check expiration date of filter paper
- Fill in all the demographic areas
- Fill in primary care physician that will assume responsibility for infant after discharge
- Obtain a good blood spot

Are you in need of newborn screening filter paper forms?

- Contact Lab Supply at (803) 896-0913

NOTE: Please do not delay mailing your specimens to the DHEC lab. There is someone to receive specimens daily regardless of lab closings.

If your office or hospital doesn't collect the repeat newborn screen please schedule a collection at your local health department:

Low Country Region: (803) 759-3000

Pee Dee Region: (843) 673-6562

Midlands Region: (803) 635-6481

Upstate Region: Ask for the lab department.

- Anderson: (864) 260-5541
- Greenville: (864) 282-4100
- Spartanburg: (864) 596-2227

ON THE SPOT

The hospitals listed below had 0% unsatisfactory specimens for the 4th quarter:

- Georgetown Memorial Hospital
- Kershaw Health

CONTACT US. WE'RE HERE TO HELP!

DHEC Newborn Screening Follow Up: (803) 898-0593/(803) 898-1969

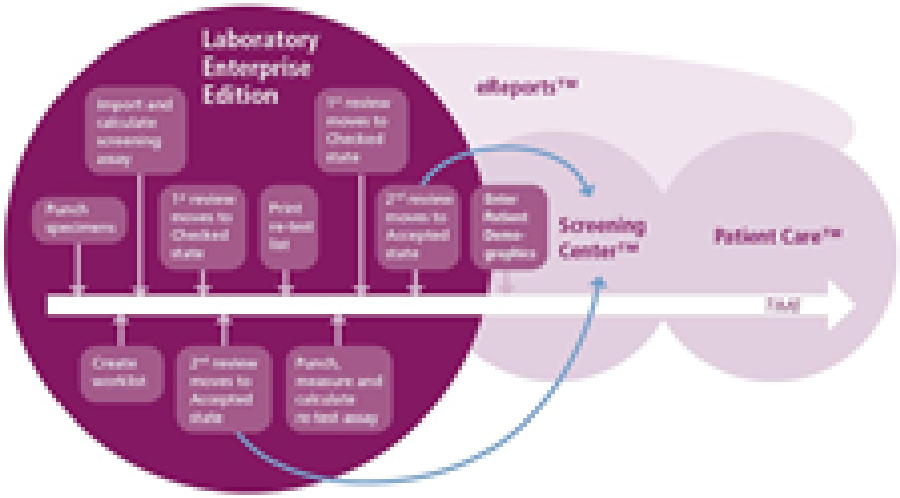
DHEC Lab: (803) 896-0891

Keep us on our toes. Please give us feedback on what you would like to see in our next Footnotes Edition. Email newbornscreening@dhec.sc.gov with your suggestions.

Are you educating your parents about Newborn Screening? Visit our website at **www.scdhec.gov/newbornmetabolicscreening** to find our newborn screening brochure and educational handouts for parents and providers.

CR-011893 2/17

On the Horizon: Laboratory Data System



Specimen Gate® Data Flow

Laboratory Enterprise Edition

- **Laboratory module** – interface with laboratory instrumentation; results management
- **Screening Center™ module** – demographic entry interface
- **eReports™ module** – secure electronic reporting web portal
- **Patient Care™ module** – interfaces with the Laboratory module and Screening Center module and facilitates newborn screening follow-up care.

http://www.perkinelmer.com/labsolutions/resources/images/for_web/SpecimenGateLaboratoryPG

Benefits to Using eReports™

- Electronic reports vs. paper reports
- Downloadable and printable laboratory reports
- 24 hour direct access to patient NBS reports
- Reduced Turn-around-time for receipt of reports

Timeline
Data System

- **Laboratory module** → go-live April 2017
- **Screening Center™ & eReports™ modules** → final phases of configuration; validation to follow
- **Patient Care™** → beginning phase of configuration

Newborn Screening Resources

Training Resources:

The training video: "*Every Hour Counts*" was produced by a national newborn screening technical assistance program in collaboration with HRSA.

Please use this link for video access:

https://youtu.be/30qbkhp1jQ8

Website Resources:

www.BabysFirstTest.org

www.newsteps.org

South Carolina Department of Health and Environmental Control
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CONTACT US

Tanya Spells, MS, MT(ASCP)
Newborn Screening Program Manager
803-898-0619

Jennifer Schlub, RDN, LD
Newborn Screening Follow Up
803-898-1969

Dana Smith, R.N.
Newborn Screening Follow Up
803-898-0593

Dr. Eileen Walsh
Pediatric Medical Consultant
Children's Health
803-898-0362

Sandi Hall, MT(ASCP)
Bureau of Laboratories
Newborn Screening Supervisor
803-896-0891

R. Brent Dixon, PhD, HCLD(ABBB), FACB
Bureau of Laboratories Director
803-896-0965

Ona Adair, PhD
Bureau of Laboratories
Chemistry Division Director
803-896-0991

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